Course description

Part 1

General information about the course	
1. Major of study: Nursing	**2. Study level:** First degree **3. Form of study:** Stationary
4. Year: I/2021-2026	**5. Semester: I**
6. Course name: Genetics	
7. Course status: obligatory	

8. Course contents and assigned learning outcomes included in the standards

Providing knowledge about the function of the genome, transcriptome and human proteome.

Providing knowledge about the basic concepts of gene expression regulation, including epigenetic regulation.

Providing knowledge about the structure of chromosomes and the molecular basis of mutagenesis.

Providing knowledge of the principles of inheriting different numbers of traits, inheriting quantitative traits, independent inheritance of traits, and inheritance of non-nuclear genetic information.

Developing skills to estimate the risk of disclosure of a given disease based on the principles of inheritance and the impact of environmental factors.

Developing skills to use knowledge about genetically determined diseases in cancer prevention and prenatal diagnosis.

Learning outcomes / reference to learning outcomes indicated in the standards
For knowledge – student knows and understands: C.W9, C.W10, C.W11, C.W12,
For skills student can do: C.U3, C.U4

9. Number of hours for the course	**40**
10. Number of ECTS points for the course	**1**

11. Methods of verification and evaluation of learning outcomes

Learning outcomes	Methods of verification	Methods of evaluation*
Knowledge	One- choice test	*
Skills	One- choice test	*

* The following evaluation system has been assumed:

Very good (5,0) – the assumed learning outcomes have been achieved and significantly exceed the required level
Better than good (4,5) – the assumed learning outcomes have been achieved and slightly exceed the required level
Good (4,0) – the assumed learning outcomes have been achieved at the required level
Better than satisfactory (3,5) – the assumed learning outcomes have been achieved at the average required level
Satisfactory (3,0) – the assumed learning outcomes have been achieved at the minimum required level
Unstatisfactory (2,0) – the assumed learning outcomes have not been achieved

Course description

Part 2

Other useful information about the course	
12. Name of Department, mailing address, e-mail: Department of Biochemistry and Medical Genetics, 40-752 Katowice Medyków str. 18, phone 32 252 88 64, biogen@sum.edu.pl	
13. Name of the course coordinator: PhD Paweł Niemiec prof. SUM	
14. Prerequisites for knowledge, skills and other competencies: Basic knowledge about genetics and biology.	

15. Number of students in groups	In accordance with the Senate Resolution
16. Study materials	http://biochigen.sum.edu.pl, notice board of Department of Biochemistry and Medical Genetics
17. Location of classes	Department of Biochemistry and Medical Genetics, workroom no 10, building C1, Medyków 18, Lecture room of School of Health Sciences
18. Location and time for contact hours	http://biochigen.sum.edu.pl

19. Learning outcomes

Number of the course learning outcome	Course learning outcomes	Reference to learning outcomes indicated in the standards
C_K01	Functions of the genome, transcriptome and human proteome.	A.W10
C_K02	Basic concepts of gene expression regulation, including epigenetic regulation.	A.W10
C_K03	The structure of chromosomes and the molecular basis of mutagenesis.	A.W11
C_K04	Basic rules of inheritance.	A.W12
C_K05	Genetic determinants of human blood groups and serological conflict in the Rh system	A.W9
C_S01	Estimate the risk of exposure of the disease based on inheritance rules and the impact of environmental factors.	A.U3
C_S02	Use knowledge about genetically determined diseases in cancer prevention and prenatal diagnosis.	A.U4

20. Forms and topics of classes	**Number of hours**
20.1. Lectures	**30**
Structure and function of genetic material - basic genetic concepts; structure of DNA, RNA, chromatin, gene, genome; the mitochondrial genome; cell cycle and replication - basic assumptions; genetic code and expression of genetic information - basic assumptions.	**3**
Variation and heredity - Hereditary variability: recombination and mutational. Molecular basis of mutagenesis - formation of single-gene and chromosomal mutations. Spontaneous	**3**

and induced mutations. Mutagenic factors - physical, chemical, biological. Repair of mutations and DNA damage.	
Mechanisms of epigenetic inheritance - The main mechanisms of epigenetic control of gene expression - DNA methylation, histone acetylation, RNAi (miRNA, dsRNA). Disturbance of the epigenetic profile and diseases. Factors causing epigenetic changes. Parental imprinting. Characteristics of X chromosome inactivation - the role of the XIST gene and its methylation.	**3**
Basic principles of inheritance - single gene inheritance. Features of autosomal dominant and recessive inheritance. Features of X-linked dominant and recessive inheritance. Incomplete dominance, codomination, multiple alleles. Examples of autosomal dominant inherited diseases (achondroplasia, myotonic dystrophy, Marfan's syndrome, Huntington's disease, osteogenesis imperfecta) and recessive (cystic fibrosis, sickle cell anemia, monogenic metabolic blocks - tyrosinemia, phenylketonuria, alkaptonuria, albinism). Examples of X-linked diseases, recessive (Duchenne and Becker muscular dystrophy) and dominant (hypophosphatemic rickets types I and II, fragile X chromosome syndrome).	**3**
Basic principles of inheritance - multi-gene inheritance. The interaction of many genes in conditioning one trait of a cumulative, complementary and epistatic nature. Interactions between genetic and environmental factors in determining phenotype. Opportunity, odds ratio, risk, synergy. Examples of multigenic and multifactorial diseases: coronary artery disease, diabetes mellitus type I and II, arterial hypertension, mental, autoimmune and neurodegenerative diseases.	**3**
Genetics and cancer prevention. The basis of neoplastic diseases: proto-oncogenes, suppressor genes, repair factors. Models of tumor formation: two-hit and multi-hit models. The most common hereditary neoplasms: breast and ovarian cancer (mutations of BRCA1 and BRCA2 genes), colorectal cancer (mutations of MLH1, APC genes). TP53 gene and Li-Fraumeni syndrome. DNA repair deficits - Xeroderma pigmentosum.	**3**
Principles of genetic counseling - part 1. Conditions determining the validity of genetic counseling. Elements of genetic counseling. Cytogenetic methods and molecular biology techniques used in the diagnosis of genetic diseases.	**3**
Principles of genetic counseling - part 2. Prenatal diagnosis - non-invasive methods (USG, Doppler examination) and invasive methods (chorionic villus sampling, amniocentesis, cordocentesis, fetoscopy). Genetic preimplantation diagnostics. Gene therapy.	**3**
Personalized medicine. Traditional approach and personalized medicine. Factors influencing the effectiveness of traditional therapies. Molecular stratification of patients. Pharmacogenetics and pharmacogenomics. Examples of the use of personalized medicine in oncology, cardiology and other fields of medicine.	**3**
Elements of population genetics in the context of human diseases. Genetic diversity of the human population, differences in the occurrence of genetic diseases, genetic polymorphism. Hardy-Weinberg equilibrium. Factors influencing the frequencies of genotypes and alleles in the population: selection, mutations, isolation, migration, genetic drift. Genetic burden of the population. Eugenics.	**3**
20.2 Classes	**10**
Estimating the risk of genetic diseases. Inheritance of monogenic diseases (autosomal recessive, autosomal dominant, X-linked recessive, X-linked dominant) - pedigree analysis, single- and several-gene crosses. Population risk. The role of environmental factors in conditioning single-gene diseases (phenylketonuria, hyperhomocysteinemia). Genetic determinants of blood groups and causes of serological conflict in the Rh system - crossbreeds. Non-nuclear inheritance.	**5**
Multi-gene and multi-factor inheritance. Gene interaction: cumulative genes, complement,	

epistasis. **Dysmorphological diagnosis of genetic diseases.** Structural and numerical chromosomal aberrations. Dysmorphic features in the most common chromosomal syndromes (Down, Klinefelter, Turner, Edwards, Patau syndrome), microdeletion syndromes (Cr idu chat, Prader-Willi, Angelman, Williams, Wolf-Hirschhorn syndrome) and monogenic diseases (Marfan syndrome, Achondroplasia, fragile X chromosome syndrome).	**5**

24. Readings

1. Alberts B et al. Molecular biology of the cell. New York: Garland Science, 2008.
2. Jorde LB et al. Medical Genetics. Elsevier, 2015.
3. Epstein RJ. Human Molecular Biology. Cambridge: Cambridge University Press, 2003.
4. Connor M., Ferguson-Smith M. Essential Medical Genetics. Wiley-Blackwell, 1997.

25. Detail evaluation criteria

In accordance with the recommendations of the inspection bodies
Completion of the course – student has achieved the assumed learning outcomes
Detail criteria for completion and evaluation of the course are specified in the course regulations